

## **Families of people with Marfan syndrome. The relationship between the functioning of the family system and the life quality of the affected persons**

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### **Summary**

**Aim.** The purpose of the study was to analyze the functioning of a family as perceived by a person with Marfan syndrome and to look for relationships between the characteristics of the system and the overall quality of life of the ill.

**Material and method.** Participants included 33 individuals with Marfan syndrome and 33 individuals without chronic illness. We used the *Family Evaluation Scale* – the Polish adaptation of the FACES-IV by D.H. Olson and the *Satisfaction with Life Scale* (SWLS) by Diener, Emmons, Larson and Griffin.

**Results.** People with Marfan syndrome perceive their families as significantly less coherent and significantly more disengaged than people without chronic illness. This family system of people with Marfan syndrome can be characterized by low scores on the “Cohesion” and “Flexibility” and high scores on the other four scales showing the level of imbalance of the family as a system, which makes this family profile similar to an ‘unbalanced’ system. Life satisfaction of people with Marfan syndrome correlated positively with “Cohesion”, “Flexibility” and “Family Satisfaction” as features of the family system perceived by them.

**Conclusions.** The obtained results confirm the importance of supporting families of people with Marfan syndrome and specialist help aimed at dealing with emotional burden related to the health of the patient.

**Key words:** Marfan syndrome, family system, cohesion, flexibility, quality of life

## Introduction

Currently, the biomedical model is the dominant approach towards patients. The special role of psychologists is to show the possible ways to support somatically ill patients and their families. So far, psychotherapy was associated with support for people suffering from mental disorders. The scientific investigation of the psychosocial functioning of people with Marfan syndrome will allow to direct professional support as well as to identify areas where help is needed.

Studies on the psychosocial and family functioning of people with Marfan syndrome show another area in which increasing knowledge and suggesting new directions of specialized influence can help improve the quality of life. Emphasizing the role and the potential of psychotherapy in the service of medicine is especially important in situations where the specialist has a specifically defined framework of helping the patient. Multidimensional studies on the chronically ill contribute to the development of the biopsychosocial approach [1].

As for the issues related to the family system's response to a family member's illness, only a small number of studies were conducted, and those that were carried out using the Family Systems Theory approach largely concern cancer patients [1].

There is insufficient research on the psychosocial aspects of the lives of people with Marfan syndrome [2]. Past research shows that people with Marfan syndrome have a lower quality of life, that they face challenges in education, work, or family life, and that they suffer from depressive and anxiety disorders. So far, no studies have been conducted on the families and the functioning of family systems of people with Marfan syndrome [2].

### Marfan syndrome — medical and psychological aspects

Marfan syndrome is a genetic disorder of the connective tissue, caused by a mutation in the fibrillin-1 gene [3, 4]. This syndrome has a diverse clinical picture and involves multiple organs and systems. Skeletal abnormalities are the most recognizable characteristic: excessive height, deformed chest, spine curvature disorders, disproportionally long — compared to the torso — upper and lower limbs, elongated fingers and toes (so-called: arachnodactyly) and facial dysmorphism [5-7]. Patients with Marfan syndrome are often affected by serious changes in their visual system, most often severe myopia and ectopia lentis, which is a displacement of the eye's crystalline lens into a non-anatomic position [8]. However, the most serious abnormalities occur in the cardiovascular system and they are responsible for significantly reducing the life expectancy of patients with Marfan syndrome. The most common causes of death among patients with Marfan syndrome are ruptures and dissections of aortic aneurysms [9, 10].

The clinical course of Marfan syndrome has a significant impact on the mental state of the patients. The awareness of the risk of death due to the rupture or dissection of aortic aneurysms, the need to undergo cardiac surgery, or the increase of sight abnormalities and the risk of loss of vision are a heavy burden. Moreover, in the case

of simultaneous occurrence of Marfan syndrome in the family, the patients often experience the sudden death of a close family member. Marfan syndrome also causes numerous limitations in patients' everyday life; those are the result of perceived ailments such as the feeling of chronic fatigue or pains in joints, the inability to exercise, the need to avoid injury, or strict adherence to medical recommendations, for example, after implantation of a mechanical valve. Moreover, their distinct appearance (extreme height, weight deficiency, disproportionally long limbs) is linked to difficulties in social functioning, as well as to stigmatization and rejection.

Studies show that people with Marfan syndrome have a significantly lower quality of life [11-13]. There is a higher than average level of psychological stress and an increased risk of mental illness among those patients [13]. People with Marfan syndrome, due to their health issues, have problems getting hired and they are subjected to workplace discrimination. Life-threatening complications, their characteristic appearance and the stigmatization they face can lead to emotional disorders [2]. The analysis of the available research results shows that psychopathological symptoms are more common among people with Marfan syndrome than in the general population. The occurrence of depression and anxiety disorders was especially apparent [12, 14]. The increased fatigue has an effect on their well-being and mental functioning. In addition, the illness is associated with physical limitations, due to the abnormal functioning of the circulatory and respiratory systems as well as due to decreased visual acuity [11]. Physical limitations make it difficult to perform daily activities, such as going to school or work, playing sports or participating in social life.

As a defense reaction in the face of life-threatening possibilities, defense mechanisms — especially repression — start to operate [15]. The social behavior of adults stems from the way they were treated in childhood. The mocking and teasing of children with Marfan syndrome due to their different appearance causes them to be withdrawn and to experience difficulties connecting with others [16]. Authors of other studies show that the distorted body image of people with Marfan syndrome negatively influences their sex life [2]. In addition, the well-being of patients is also influenced by the medication they take, and the surgeries and hospitalizations that they experience [11].

Receiving a diagnosis for a life-threatening disease such as Marfan syndrome is associated with anxiety [17]. The process of receiving the diagnosis and giving the information to the patient is conditioned socially and culturally, and surrounded by numerous controversies. Giving the news to the patient and their family influences the course of the process of dealing with the disease not only by the patient, but also by the family. Regarding the delivery of the diagnosis, there are two coexisting models, existing on two extreme spectrums: a model of open communication about the diagnosis and prognosis, and a model that postulates keeping the diagnosis and prognosis a secret, which results in a „conspiracy of silence”. The latter approach is more widespread in Poland and one of its consequences is the lack of communication with one's family and friends about the illness and the emotions that accompany it. This occurs in the setting of a fictitious atmosphere, imbued with lies. Consequently, this leads to the weakening of the bonds and emotional distancing of the members of the patient's family. The emotions of the patient are suppressed, which in a longer

perspective can cause the loss of emotional control. On the other hand, the opposite approach takes away the person's hope and undoubtedly has a negative impact on their mental health and as a consequence, on their physical health [17-19]. The anxiety that appears in the family system, associated with the risk of losing a loved one, can either help bring people closer together, or — by avoiding the difficult topics and refraining from sharing emotions — drive a wedge between the family members, depending on the strength and the quality of the relationship.

### **Family with a chronically ill person**

Family is our natural and primary environment. The Family Systems Theory paradigm perceives the family as an integral whole — a multidimensional phenomenon with a structure, a system of roles, power and control, as well as communication schemes. In the Family Systems Theory, every change in the family or affecting individual members is perceived as the result of the functioning of the system. The proper functioning of the family system provides it with the ability to maintain homeostasis, that is, to maintain stability. The fundamental assumption is that a change in one part of the system changes the system as a whole, and at the same time, a lack of involvement to change even just one element of the system, blocks the process of change in the whole system [20-22].

In the life cycle of a family, the system is mobilized to change multiple times as a result of events that affect its stability. The family usually moves smoothly through subsequent phases of family life caused by such events as the birth of a child, growing up, or becoming independent [23]. There are also other factors that provoke a change in the family system, and those are sudden and unexpected life events, such as sickness of one of the family members, described by Carter and McGoldrick [23] as unpredictable horizontal stressors. How the family will deal with them may depend on vertical stressors, that is, rules, principles and beliefs, transmitted from generation to generation, concerning, among other things, sickness or the role of the sick person in the family. Chronic illness disrupts the achievement of goals and life tasks, and its influence on the individual leads to the consolidation of those changes that were the primary, reactive and temporary response of the family system to change. Changes in the family caused by illness include, but are not limited to: disturbances of the ordinary flow of the day, financial troubles, difficulties carrying out long – and short-term plans, loss of physical closeness between the spouses, conflicts and communication issues in the family, isolation from external matters and concentration on the family problems — which can cause loss of flexibility or closing of the boundaries of the family system [24].

Due to a large number of negative consequences for the patients and their families, each illness is evaluated negatively — as a stressor to all of its members [25]. There is a relationship between the course of the disease and the intra-family processes. The importance that a family places on the illness modifies the reactions of the family. Perceiving the illness as a serious threat usually causes anxiety and depression, while perceiving the illness as a challenge, mobilizes energy and resources needed to

deal with it [1]. The adaptation of the person to the situation of worsening health and the attitude of the family regarding the process of dealing with the family member's illness can be influenced by such factors as the schema of the illness, understood as the patient's set of beliefs and feelings towards the illness, based on the information obtained from the doctor and other sources of information and modified by the patient's own interpretation [25]. It is the key to understanding the emotional and behavioral relation to illness [1].

Rolland [26] formulated the assumptions of the family system approach in the face of illness of one of the members. He suggests that when trying to understand the situation of chronically ill patients and their families, the illness should be perceived as a multilevel illness system. This model takes into account three dimensions. The first one is psychosocial aspects of the illness, which indicates that the type of illness needs to be taken into account when designing the support for the family. In this area, Rolland distinguishes fatal illness, illness that shortens the life span, and illness that is not fatal. Another level in the psychosocial dimension of the illness is its course, analyzed through questions on whether the illness is progressive, whether there are recurrences, or whether the illness does not progress with time. Studies are also being conducted on the intensification of disability, since the illness can cause sudden disability or the person can become increasingly disabled as the illness progresses, bringing on further losses.

Conceptualizing the psychosocial aspect of a chronic illness, as a second dimension of the *Family Systems-Illness Model*, Rolland [26, 27] takes into account the life cycle of a person, a family and an illness. In this dimension, the illness is perceived as a process split into a phase of crisis, a chronic phase and a terminal phase. At each stage, the family has a different developmental task and the role of each member is to adapt to each of the stages. The third dimension is the transgenerational history of illness, referring to the losses and the crises of each particular family, which includes a system of beliefs, culture and the traditions cultivated by the family. This dimension underlines the importance of extending the family history in the systematic perspective with aspects of dealing with illnesses in the past, such as adaptation schemes, repetitions, lack of continuity, or changes in family relations (alienation, triangulation, separations). Such schemes are transmitted throughout generations as family mythology, taboos, catastrophic predictions and belief systems [28].

Understanding the progression of the illness, the individual development of family members, including the ill person, and the developmental cycle of the family as three intermingled threads is a very complex process that has been insufficiently explored in the case of people with Marfan syndrome. Conducting research in the transgenerational paradigm seems particularly important due to the particularity of the Marfan syndrome, where 75% of the ill inherited the disorder [13]. Therefore, the family has a history of sudden losses and numerous convictions, such as those regarding the possibility of saving a loved one's life.

## Characteristics of the family system and quality of life of people with Marfan syndrome

Previous studies do not provide a clear answer regarding the impact of the illness on the family system. Results implicate strong reactions of the system to the illness of a family member, which are aimed at maintaining balance to ensure a sense of security. Activation of mechanisms such as denial of conflicts, limitation of communication and suppression of emotions [1, 29] are often noted. Thus, the results of research conducted so far implicate the need for further exploration.

One group of researchers argues that an illness of a family member causes the family to get closer to one another and that an illness is an integrating factor [1, 20, 25]. In families in which the bond was strong even before the diagnosis, a serious illness in the family caused the family members to get closer to one another and to intensify the mutual support. However, a second group of researchers showed that in a situation of long-term illness, the intra-family boundaries become blurred and the external boundaries stiffen. They also showed the reactions of family members, which included hostility towards one another, rejection of the ill family member, demonstrating to members of other families the tragedy of the situation, and expressing a lack of faith in the treatment. This was especially visible among the families who had been previously in conflict or who only appeared to have a good relationship [1, 24, 29-31]. It was also destructive to over-focus on the illness — that is, for the family to wrap itself around the illness — as it blocks the family from taking on subsequent developmental tasks [1], linked to, for example, worsening of the health of the patient and to necessary inclusion of a greater number of people in order to take care of the ill person.

Previous research on the quality of life of people with Marfan syndrome was mostly concerned with the quality of life linked to physical health. Those results suggest that adults with Marfan syndrome experience a reduced overall quality of life, although a more careful analysis shows some discrepancies and ambiguities. One of the studies showed a reduced quality of life depending on the health (HRQoL) in all of the subscales of the questionnaire compared to the general population of Norway, while at the same time not showing any relationship between the medical criteria of the Marfan syndrome and the health depending quality of life (HRQoL) [11]. Studies conducted in the US show a reduced quality of life (HRQoL) only in the area of „physical activity”, compared to the general population [32, 33]. Other research conducted with the use of the *Quality of Life Index-Cardiac Version III (QLI)* showed lower scores in the area of psychological functioning among people with Marfan syndrome compared to the population of people suffering from other cardiological conditions [12].

### Research purpose

The purpose of the study was to analyze the functioning of a family as perceived by a person with Marfan syndrome and to look for relationships between the characteristics of the system and the overall quality of life of the ill. We created three detailed goals: 1) to define the differences between people with Marfan syndrome and

people without chronic illness regarding their perception of family functioning; 2) to define the profile of the family system of people with Marfan syndrome; 3) to define the relationship between the family system characteristics and the quality of life of people with the disorder. The study was carried out as a part of the BST2016 project of the SWPS University of Social Sciences and Humanities, Faculty of Psychology in Sopot (WSO/2016/A/04) regarding the Marfan syndrome as a rare genetic disorder in the context of evaluation of quality of life and personal resources. As a part of a larger research project on families, it received a positive opinion from the Research Projects Ethics Committee at the Psychology Institute of the University of Gdansk (request number 3/2016).

Detailed research questions were proposed:

1. Are there differences between people with Marfan syndrome and people who are not chronically ill in terms of perception of the family system characteristics and the level of family balance?
2. What is the profile of a family system of a person with Marfan syndrome?
3. Is the functioning of a family linked to the level of quality of life of a person with Marfan syndrome?

Although there are premises in literature that allow us to form assumptions regarding the functioning of families of chronically ill people [1], due to the lack of reports on the families of people with Marfan syndrome and the small amount of research on the psychological functioning of ill people, it is more difficult to propose the direction of hypotheses. In connection with this, it has been assumed that there exist differences between people with Marfan syndrome and people not chronically ill, in terms of perception of the family system and the level of family balance. These differences will particularly concern the area of family ties, which will be expressed by a different level of cohesion of the family system. No hypothesis was proposed for the second question due to lack of research on the functioning of family systems. In this area, own research will be exploratory in nature. It was assumed, however, that there is a relationship between the functioning of the family system and the quality of life among people with Marfan syndrome. The higher the level of system features indicating a family unbalance, the lower will be the quality of life of patients.

## Participants

Participants included 66 people — 33 with Marfan syndrome and 33 without chronic illness. Both groups included 21 women and 12 men. Mean age was 33 years, both in the criterion ( $M = 32.68$ ;  $SD = 11.97$ ) and the comparison group ( $M = 33.03$ ;  $SD = 11.95$ ). Most participants in both groups had high school education (among people with Marfan syndrome — 16 people, among healthy participants — 11) or higher education (respectively 15 and 13 participants). The majority of the participants with Marfan syndrome were in a relationship (married — 12 or informal — 8 people), one person was divorced, and 12 people did not have a partner. In the group without a chronic disease — 21 people were in a relationship (formal — 13, informal — 8),



3 people were divorced, and 9 never had their own family. Some of the participants had children — 13 people with Marfan syndrome and 17 without chronic illness. This indicates that the compared groups were equivalent in terms of age, education and family situation (relationships). Slightly more participants without chronic illness had children, which can be a consequence of their health status and the life situation of a person with a chronic and life-threatening illness. Due to the participation of adults still living with families of origin or those who have already started their own families in own study, a family, according to the systemic approach, has been defined as a group of related people living together.

The study was conducted between 2016-2017 at the Medical University of Gdańsk, in the Department of Pediatric Cardiology and Congenital Heart Defects of the University's Medical Center. The criterion group included people who were diagnosed with Marfan syndrome or diagnosed as marfanoid patients and who agreed to participate in the study after assuring their anonymity when presenting the results. Each person was assessed individually in order to obtain medical and psychological history, fill out the questionnaires and was given the opportunity to ask any additional questions. Persons in the criterion group, due to the ongoing diagnostic process, were not subjected to pharmacological therapy dedicated to the disease. From the perspective of Rolland's systemic model of family with illness [26, 27], due to the diagnosis of certain symptoms of the disease and the simultaneous uncertainty as to the diagnosis and hope for the lack of confirmation of the diagnosis, the families of the respondents were in a crisis phase.

### Materials and procedure

To evaluate the family system and the family profile, we used the *Family Evaluation Scale*, which is the Polish version of FACES-IV by D. H. Olson in adaptation by Margasiński [34, 35]. The questionnaire has eight subscales, split into two subgroups: 1) six main scales of the *Circumplex Model*: Balanced Cohesion, Balanced Flexibility, Disengaged, Enmeshed, Rigid and Chaotic and 2) two evaluating scales — Family Communication and Family Satisfaction. The tool consists of 62 items that the participants use to evaluate the degree of their agreement on a 5-point scale where 1 means „I totally disagree” and 5 — „I totally agree”. The questionnaire has high or satisfactory internal validity of the subscales, and the confirmation analysis showed a structure consistent with the *Circumplex Model* [35]. In this study, the reliability analysis showed the following values of Alpha Cronbach for each subscale: Balanced Cohesion — 0.756, Balanced Flexibility — 0.670, Disengaged — 0.818, Enmeshed — 0.625, Rigid — 0.6, Chaotic — 0.655, Communication — 0.908 and Family Satisfaction — 0.908.

The *Satisfaction with Life Scale* by Diener, Emmons, Larson and Griffin, adapted to Polish by Juczynski [36], is used to measure the perceived satisfaction with life. It consists of five items, evaluated on a 7-point scale, from „strongly agree” (7 points) to „strongly disagree” (1 point). The respondents are answering to what extent each of the statements relates to their life. The higher the result, the higher the satisfaction with life. In this study, the scale's reliability was  $\alpha = 0.827$ .



## Results

In order to answer the first research question, regarding the differences between people with Marfan syndrome and people who are not chronically ill, concerning the perception of the family system's traits and the degree of family balance, we analyzed the data using the *t*-Student test. The results are presented in Table 1 and in Figure 1. We found significant differences between the criterion and comparison groups only for the subscales concerning the strength of family ties. People with Marfan syndrome perceive their families as significantly less coherent and significantly more disengaged than people without chronic illness ( $p < 0.05$ ). The *d*-Cohen index has medium values.

Table 1. Significance of differences regarding the family system's traits between participants with Marfan syndrome and participants without chronic illness

	Participants with Marfan syndrome		Participants without chronic illness		t	df	p	Cohen's d
	M	SD	M	SD				
Balanced Cohesion	26.09	4.35	28.40	4.83	-2.016	63	.048	-0.50
Balanced Flexibility	22.22	4.19	23.88	4.50	-1.448	63	.153	-0.25
Disengaged	16.38	5.66	13.18	4.92	2.431	63	.018	0.50
Enmeshed	15.13	4.20	13.70	4.52	1.319	63	.192	0.25
Rigid	17.38	4.27	16.85	4.19	.502	63	.618	ns
Chaotic	17.81	3.98	17.40	5.29	.360	63	.720	ns

In the remaining scales of family evaluation as well as in the case of satisfaction with family life (Table 2), the results suggested no differences between the groups in terms of perception of family functioning as a system.

Table 2. Significance of differences in the evaluating scales between participants with Marfan syndrome and participants without chronic illness<sup>0</sup>

	Participants with Marfan syndrome		Participants without chronic illness		t	df	p	Cohen's d
	M	SD	M	SD				
Communication	35.93	7.48	39.09	7.41	-1.708	63	.093	-0.25
Family Satisfaction	36.66	6.99	39.15	7.94	-1.343	63	.184	-0.25

People with Marfan syndrome seem to be less satisfied with family communication than people without chronic disease, although for this subscale we only observed a trend ( $p < 0.093$ ), thus it is necessary to be careful when interpreting this result and to look for similar results in subsequent studies.

The *Family Evaluation Scale* allows to estimate the overall system balance indicators as well as cohesion and flexibility as the main dimensions in the *Circumplex Model* [35, 37]. The results presented below show a significant difference in the dimensions of Cohesion ( $p < 0.01$ ) and Balance ( $p < 0.05$ ) between people with Marfan syndrome

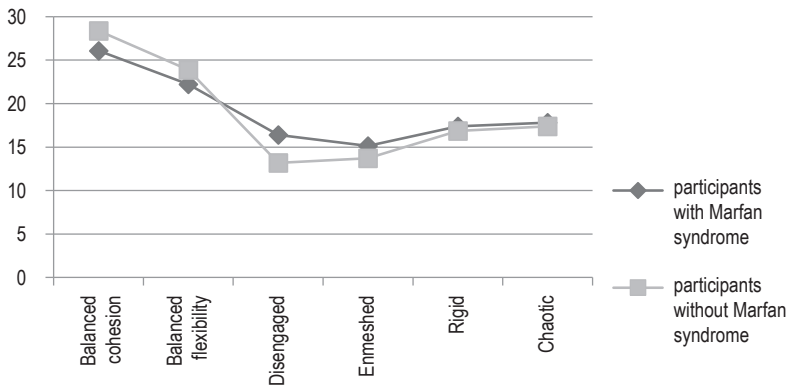


Figure 1. Results from the Circumplex Model scales among participants with Marfan syndrome and without Marfan syndrome

and people without illness. The obtained result shows that in the families of people with Marfan syndrome, their members seek relationship autonomy to a larger extent than in the families without a chronically ill person. This result is interesting, taking into consideration the theoretical models and numerous previous studies which show a tendency to maintain closeness in families with an ill person, especially in the case where there is a high risk of sudden death. On the other hand, the fear of losing a loved one might be a way to maintain individual inner balance, because closeness is linked to direct confrontation with the sick person's fear and the need to support them while at the same time experiencing anxiety. People with Marfan syndrome also perceive the level of family balance differently.

Table 3. Significance of differences of the family system balance perception between participants with Marfan syndrome and participants without chronic illness

	Participants with Marfan syndrome		Participants without chronic illness		t	df	p	Cohen's d
	M	SD	M	SD				
Cohesion index	.69	.39	1.04	.61	-2.718	63	.008	-0.50
Flexibility index	.71	.36	.85	.45	-1.352	63	.181	-0.25
Balance index	.70	.33	.93	.46	-2.305	63	.024	-0.50

The second objective of the study was to define the profile of the family system of people with Marfan syndrome. We analyzed the distribution of sten score results obtained by the participants with Marfan syndrome. In the case of Balanced Cohesion and Balanced Flexibility, as many as 93% of the participants obtained low (1-3 sten) and average scores (4-7 sten). We already described the difference of scores concerning the emotional bond between family members of persons with Marfan syndrome and the scores among people not affected by a chronic illness, thus show-

ing the particularity of the families with Marfan syndrome. The result regarding flexibility understood as a family trait that helps with changing the rules and adapting to the expectations due to the changes within the system or those affecting the family from the outside is especially interesting. Only 7% of people with Marfan syndrome evaluated their families as highly flexible, that is, as being able to adapt their functioning to the circumstances. Flexibility seems to be necessary for a family with a serious illness, as they need to act quickly in the case of a life-threatening event, such as aortic dissection.

On the Disengaged scale, 3.1% of people with Marfan syndrome obtained low scores, which means a high degree of attachment between the family members, and 40.7% obtained average scores. The remaining 56.2% of the ill perceived their families as highly disengaged, which translates to a high emotional distance between each other, which suggests, among other things, a difficulty in obtaining emotional support — which might seem obvious, and a difficulty in asking for instrumental help, for example linked to the troubles due to the illness. Low scores on the Enmeshed scale were obtained by 3.1% of people with Marfan syndrome, average scores — 71.9%, and high scores — 25%. This means that the participants from the criterion group perceive — in a moderate way — their family members as too dependent. This would manifest through spending too much time together and the felt pressure to stay close to each other and to distance themselves from people outside the family. Similar sten score profiles, with the majority of average scores, which is typical for the majority of families, were obtained on the Rigid subscale (9.4% of people with Marfan syndrome — low score, 68.7% — average score, 21.9% — high score), which means keeping the family rules no matter the outside circumstances and the situation inside the family. Similar results were obtained on the Chaotic subscale (3.1% — low score, 59.5% — average score, 37.5 — high score), which measures to what extent it is clear in the family who sets the rules and what rules apply. What is noticeable for both scales is the deviation from the normal distribution corresponding to each group of sten scores (16% — low score, 68% — average score, 16% — high score), which suggests a higher Rigid score with simultaneously higher Chaotic score. The results obtained by people with Marfan syndrome on those two subscales suggest moderate difficulties in establishing and observing rules set up by the family and also in enforcing any consequences for breaking them.

The analysis of the sten scores concerning the family perception by the participants with Marfan syndrome shows a similarity of those families' profiles to a typical profile for an „unbalanced” system [35], which is visible in the figure below (Figure 2). This system can be characterized by low scores on the two scales that relate to the level of balance in the family and high scores on the four scales which show the level of imbalance of the family as a system.

The overall (qualitative) profile analysis was further confirmed with a statistical analysis. We conducted a one sample t-Test where the tested values were the means from the „Unbalanced” profile, and the results showed that the participants with Marfan syndrome scored equally low on the following scales: Balanced Cohesion, Balanced Flexibility, and Disengaged. Despite the fact that for the other subscales

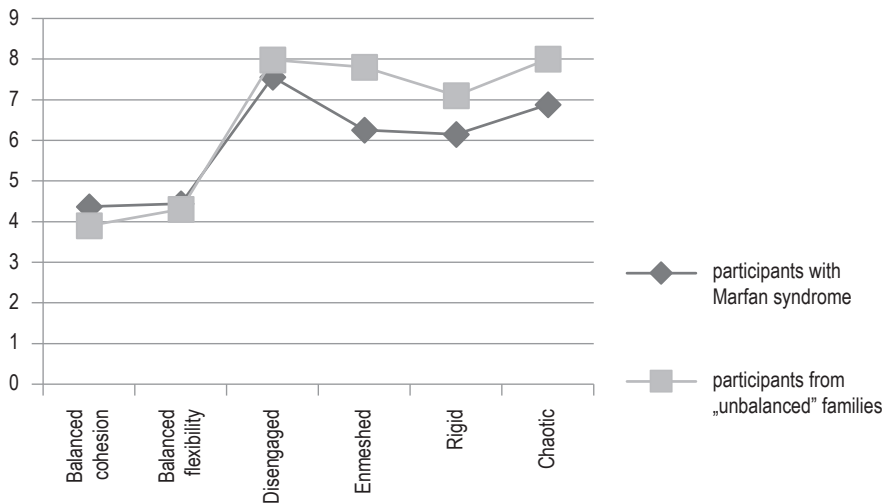


Figure 2. Comparison of the profile of participants with Marfan syndrome with the profile of „unbalanced” families

there were significant differences between the participants with Marfan syndrome and the participants in the „Unbalanced” systems, the overall profile remains similar (Table 4).

Table 4. Comparison of sten scores on the Circumplex Model scales between people with Marfan syndrome and the mean population scores

N = 32	Participants with Marfan syndrome		t-Student	df	p
	M	SD			
Balanced Cohesion	4.38	1.70	1.581	31	.124
Balanced Flexibility	4.43	1.50	.518	31	.608
Disengaged	7.56	1.81	-1.303	31	.202
Enmeshed	6.25	1.74	-5.035	31	< 0.001
Ridig	6.16	1.78	-2.999	31	.005
Chaotic	6.88	1.39	-4.594	31	< .001

Moreover, the overall family balance index showed that participants with Marfan syndrome usually perceive their families as „unbalanced”, which is characterized by high scores on all four Unbalanced scales (Disengaged, Enmeshed, Rigid, Chaotic) and low scores on the scales of Balanced Cohesion and Balanced Flexibility. In the case of as many as 85% of participants with Marfan syndrome, the family balance index was lower than 1 (on a 0-1 scale), and the closer the index is to 0, the more „unhealthy” the family system [35]. As stated by the authors of the *Circumplex Model* [37] and the

author of the Polish adaptation [35], families with such a family profile experience the most problems with functioning, compared to all the other types of families. Additionally, they have the least resources, amongst which could be strong bonds or adaptability to new situations. Not only the people with the illness, but also their close ones, that is, the whole family with the „unbalanced” profile, require professional psychological support in the form of psychoeducation regarding the illness. They also are in need of therapy, the aim of which could be to help develop the skills to support one another, express their feelings (not only the ones related to the potential loss of the ill person), and also to prepare in case they have to take action if a situation occurs where life-saving surgery is necessary.

The goal of the studies on the families affected by illness is, among others, to specify to what extent will a change in the family system (such as improving communication or getting the family members closer with each other in order help them cope better with tasks that arise in subsequent phases of the illness), be helpful for the ill, and significant for their quality of life [26, 27]. The third objective of our research was thus to define the relationship between the functioning of a family with Marfan syndrome and their life satisfaction. In the first step, we compared the overall life satisfaction scores from the criterion and comparison groups. People with Marfan syndrome scored lower ( $M = 18.73$ ,  $SD = 5.62$ ) than those without a chronic illness ( $M = 21.27$ ,  $SD = 5.73$ ); however, this difference is only a trend, which means it is not statistically significant ( $t$ -Student =  $-1.822$ ;  $df = 64$ ;  $p = 0.073$ ). The sten scores of the participants with Marfan syndrome were low (42.4% of the group) and average (42.4%). In the second step, we conducted correlation analysis, using Pearson's  $r$  test, between the family evaluation scale and overall life satisfaction among people with Marfan syndrome. Life satisfaction correlated positively with Balanced Cohesion ( $r = 0.399$ ,  $p = 0.026$ ), Balanced Flexibility ( $r = 0.357$ ,  $p = 0.049$ ) and Family Satisfaction ( $r = 0.441$ ,  $p = 0.013$ ), which means that the relationships between family members, the ability to adapt to external stressors and to the requirements that emerge as a result of family development, and also satisfaction with family functioning, are of significance for the overall life satisfaction among people with Marfan syndrome. Statistically significant coefficients are low or moderate.

## Discussion

The aim of the presented studies was to describe the functioning of a family in the eyes of people with Marfan syndrome and to link the characteristics of the family system with the overall quality of life among the ill. This is an important issue, as different tasks have to be performed by the family depending on the type of the illness, which is emphasized by, among others, Rolland in the *Family Systems-Illness Model* [26, 27]. In addition, according to the systemic way of thinking about family functioning, there is a circular interdependence between the functioning of the ill person and other members of their family. On the one hand, the health condition of the ill person influences the thoughts, feelings and behaviors of their loved ones; on the other hand, the reactions of the family members influence the well-being of the ill person, leading

to either improvement of their functioning or to worsening of the illness, depending on the provided support.

The first goal of the study was to determine the differences in the perception of the family system by people with Marfan syndrome, compared to people without chronic disease. It turned out that in the perception of the participants from the criterion group, their families had lower scores than the comparison group on the Cohesion scale and higher scores on the Disengagement scale. The hypothesis regarding the differences between people with Marfan syndrome and non-chronically ill people in terms of perceiving the features of the family system has therefore been partially positively verified, although the direction of differences was surprising. The achieved result means that emotional bonds in families of people with Marfan syndrome, from the perspective of the ill person, are weaker than in the families evaluated by people without illness. This is an interesting result, since the most common and the most often cited model of functioning of a family with illness, created by Minuchin [38, 39], posits emotional entanglement of family members, meaning strong or even symbiotic relationships in the family. Minuchin's model concerns families of people with psychosomatic illnesses, but — as evidenced by reports on psychosocial functioning of people with Marfan syndrome, also reported here — numerous symptoms are similar to those that occur in the types of illnesses that were described by the author of the Structural Family Theory. Thus, the question is, what is the reason for the result which indicates weaker than average family ties in families with a person with Marfan syndrome?

It seems that the explanation could be found in, among others, the mechanisms of coping with the anxiety of the ill persons and their loved ones. Marfan syndrome is a life-threatening condition [26], where death can occur suddenly, especially when there is no adequate medical care and no preventive measures have been put in place. This is linked to the great concern of the ill person and the members of their family. In direct contact during the study, the participants often said that they „live on a bomb”, or „with a ticking bomb”. The unpredictability of their health — always present despite specialist care — is difficult to live with; moreover, the unpredictability is increased as there are no physical signs linked to the cardiovascular symptoms, which are closely related to the risk of death [10]. The anxiety, which accompanies the people with an illness that threatens their lives, activates defense mechanisms in the functioning of the person and their family, such as denial and suppressing emotions [17, 40]. Maryniak and colleagues [15] noted the presence of internalization disorders (anxiety, depression, withdrawal) and defense mechanisms that appear among people with Marfan syndrome. Clamming up of the ill people, their solitary distress and limited communication with their families, makes the families unable to understand what is going on with the ill person and how they can be helpful. Their lack of instrumental and emotional support can be perceived by the person with Marfan syndrome as a lack of interest and concern, which contributes to the secondary deterioration of the family relations.

The distancing of the family members of a person with Marfan syndrome can also happen because this illness is uncommon. As such, access to knowledge about

the illness and the possible treatment and rehabilitation plans is limited, which causes more anxiety and — in the face of helplessness — the tendency of family members to emotionally withdraw from the relationship. This is why different initiatives that bring ill people together, through internet fora or associations (such as Polish Marfan Association, or Association of Families of People with Marfan Syndrome and Other Genetically Determined Disorders) are so important, as they not only provide information through brochures and organizing conferences, but also function as support groups. What is symptomatic and what confirms the difficulty in making peace with the diagnosis and accepting the health issues, are the statements of the patients, which show resistance and fear related to accepting this kind of help because they need to confront their anxiety. Seeing the obtained results, it is justified to postulate that whole families should participate in the initiatives organized by the associations that operate for people with Marfan syndrome, which — apart from the benefits of acquiring information — fosters integration of the family system of the sick person [41].

The second objective of the study was to determine the profile of families of people with Marfan syndrome in terms of the family system balance. It turned out that people from the criterion group perceive their families as „unbalanced”, which means they have low cohesion and low flexibility scores [25]. This means that it is hard for those families to respond to situations that require the implementation of sudden changes, which is an important task of the family system of a person with a life-threatening illness [26, 27]. Thus, people with Marfan syndrome perceive their families as requiring support and therapy, which is typical for families of people with chronic illness [42, 43]. As mentioned above, low cohesion can serve to protect against the fear of death experienced by the ill person and their loved ones [15] and to repress the thoughts about the illness [41]. If a family is coherent and flexible (reacts to changes and challenges related to the course of the disease or a sudden need of medical intervention), and open to help from people outside the family system, then the burden for each family member is reduced, which helps the system to calm down and to provide more effective support for the patient [41].

The third research objective was to determine the level of life satisfaction and its links to family functioning. The obtained result shows only a statistical tendency in terms of differences in life satisfaction of people with Marfan syndrome and people who are not ill. It is difficult to clearly state why this result is not consistent with previous reports, in which the same tool was used (SWLS), which showed a lower quality of life among the ill [2]. Other reports indicating a lower than average quality of life concerned health aspects [13] and the difference between their results and the results we obtained might be due to the activation of defense mechanisms that help to maintain the overall satisfaction with life. Moreover, as this study shows, the stronger the family bonds, the ability to adapt to change and satisfaction with family life, the more the overall life satisfaction of people with Marfan syndrome. The hypothesis to the third question has been positively verified. The obtained results confirm the importance of supporting the family as a whole and specialistic help aimed at dealing with emotional burden related to the health of the loved one and the tasks that follow



(contact with doctors, constant monitoring of health, personal load regulation, etc.) for the ill person's functioning. It is important to remember that the analyzed variables are interdependent, and thus it is possible that people with a high overall life satisfaction tend to evaluate their families more positively in the categories of cohesion, flexibility and family satisfaction.

Acceptance of the illness and living with it depends mostly on the subjective perception of the severity of the illness and a lower number of reported symptoms by the patients [17, 44]. Thus, it is important to work on a positive attitude of the ill person towards the illness [45]. However, perceiving one's illness as severe and as hard to treat intensifies the anxiety of the patients and their families [17]. Psychological support should therefore concern not only the individual and be targeted at coping with tensions related to health and the planned course of treatment, but it should also concern the whole family of the ill person. The purpose of the support should be not only to show how the functioning of the ill persons and their loved ones is related, but also to shape communication skills and skills that help nurture family ties when facing challenges such as this illness.

It is worth underlining that the present study contributes new information not only to the local researchers, but also to global research. The only studies conducted so far that analyzed families with Marfan syndrome concerned the decision of the ill persons to marry or to be in a relationship and whether to have children [2, 46], due to the possibility of transferring the illness to the children, and also due to the burden on one's health, including the risk of death during labor [47, 48]. This study included both people with diagnosed Marfan syndrome, as well as patients with Marfan-like phenotypes which — the low sample size aside — can be treated as a limitation of the study, as was noted by the authors of a meta-analysis of psychosocial aspects of living with the Marfan syndrome [2]. On the other hand, the specialists emphasize that both illnesses are characterized by symptoms related to the skeletal, cardiovascular, visual and central nervous system dysfunctions [49], which can have similar consequences for the mental and social life of the ill person. Differential diagnosis is important not only because of the need to choose the appropriate course of treatment, but also because it is another type of life-threatening illness [26], which is of great psychological importance. Having Marfan-like phenotype is less life-threatening than having Marfan syndrome [50].

The presented report included the results of a one-time study, which was conducted in the process of patient diagnosis. Further studies should be conducted taking into consideration the process of change in the family system in a long-term perspective. This would make it possible to track the changes in the family, including coping with the diagnosis, with the progression of the illness, and also with the involvement of the person with Marfan syndrome in complying with the medical recommendations in preparation for necessary surgery. Longitudinal studies could help verify the hypothesis that the functioning of the systems with limited resources worsens over time, while the bond gets stronger between the members of families that function correctly after the chronic illness diagnosis [1]. It would also be worthwhile to — in addition to increasing the number of the participants with diagnosed

Marfan syndrome — study whole family systems, so as to consider all of the family members' perceptions. Another direction of research that emerges as a need in the face of the results presented in the report is comparison of the functioning of families of a person with Marfan syndrome with the functioning of systems in which other chronic diseases are present.

This measure would allow to uncover the features and mechanisms present in family systems that are common in a variety of chronic diseases as well as those that are specific to families of a person with Marfan syndrome.

### Conclusions

1. Families of people with Marfan syndrome are characterized by lower cohesion and higher disengagement than families in the comparison group. Emotional bonds in the families of people with Marfan syndrome are weaker, from the patient's perspective, than in the families assessed by people not affected by illness.
2. People with Marfan syndrome perceive their families as "unbalanced" and therefore the families are characterized by low cohesion with low flexibility, which means that entire family systems of ill people may require support and/or therapy.
3. The stronger the bonds in the family, the ability to adapt to changes and satisfaction with family life, the greater the overall satisfaction with life of people with Marfan syndrome. This result additionally implies the need to implement specialist care not only of medical, but also of psychological and psychotherapeutic nature, both for the person affected by the disease and for members of the family system.

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